

Requestform Clinical Genetics Maastricht

▶ **Postal address**

Maastricht Universitair Medisch Centrum
 Laboratorium Klinische Genetica
 Postbus 5800
 6202 AZ Maastricht
 Hoofd laboratorium:
 Dr. A. van den Wijngaard
 Openingstijden laboratorium: 08:30 - 16:30
 Tel: 043-3871345 | Fax: 043-3877901
 @: cmo.klin.genetica@mumc.nl

▶ **Patient information** (* =required field)

Initials*
 Last name*
 Date of birth*
 Gender*
 Ethnical background
 Address
 Postal code
 City
 Reference / MDN
 Is patient deceased?
 Date of death

▶ **Invoice address** (* =required field)

Name*
 Address*
 E-mail
 Vat number

▶ **Referring physician** (* =required field)

Name*
 Hospital / Institute*
 Address
 Email*
 Specialism
 Department*
 Country
 Telephone
 Fax
 Copy results

▶ **Test(s) requested**▶ **Material**

Material already available in our lab?
 Date of sample collection
 Which kind of material will be included?
 DNA isolated from:

▶ **Reason of the request**

Reason of referral
 Gene involved and mutation
 Has material of family member(s) been sent previously?
 Is this an urgent request?
 Reason of urgency
 Name(s)
 Date of birth

Date of request:

Version: 01-2025

To be filled in by the lab

Datum ontvangst: Ingescande pagina's:
 Paraaf ontvangst:

monstersticker(s)

Materiaal:	EDTA	HEP	DNA	Vlok	Vruchtw.	Overig:
Aantal:						

► **Family members**

Relation to current patient

Other familial information

Will you include parental samples?

Name father

Name mother

Parental clinical details

Which kind of parental material?

Date of birth father

Date of birth mother

Reason including parental samples

► **Informed consent**

I have informed my patient that the knowledge of genetic conditions is likely to improve in the future. He/She understands that, using this knowledge, the diagnostic laboratory is able to maintain an active search to identify the genetic cause of the disease for which the test was performed.

The material and/or data of the patient may be used if the current test does not find a cause for the clinical symptoms. In that case, a follow-up investigation related to the same clinical question can be performed later without a new request. This may occur, for example, when a new variant or gene is discovered, or if a more accurate test becomes available. The original requester will be notified about this. The laboratory will not charge for this. However, please note that there may be costs for the patient from the treating physician. The patient **DOES NOT ALLOW** the material and/or data to be used for additional analysis in line with the original diagnostic request. Further use of the material has not (yet) been discussed.

► **Clinical information**

Birth weight

Current length

Age of diagnosis

Clinical relevant information

Current weight

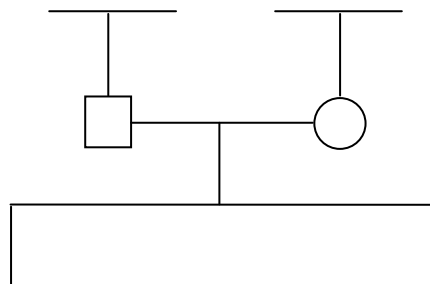
Head circumference

Consanguinity

► **Expected inheritance**

► **Pedigree chart**

Please mark the individual of this request with an arrow (→). Designate affected family members as ■ / ● and indicate previous sent family samples with name and date of birth.



► **Collaboration MUMC+**

The laboratory Genomediagnosics Nijmegen closely collaborates with the Clinical Genomics Laboratory of the MUMC+ in Maastricht. A limited part of the test offer (marked ¹) will be carried out by the partner laboratory in Maastricht. In that case, the diagnostic report will be issued directly by the Maastricht laboratory, but the payment will be issued by Genomediagnosics Nijmegen. Direct analyses of variants in family members detected by whole exome sequencing (WES) will be carried out by the laboratory that reported the variant originally in the index.

Material withdrawal form

▶ Patient information/sticker

Initials

Date of birth

Last name

Gender

▶ Material and shipping conditions

- Put name, gender and date of birth on each blood- or DNA tube. Improperly labelled samples will be refused. **Excess tubes will not be stored!**
- Ship samples at room temperature. **Do not freeze blood samples!**
- Do not submit material if patient has undergone bone marrow or stem cell transplant. Please contact lab.
- Upon cancellation the requested test will be charged completely.
- Additional information for requesting diagnostic testing at the Division of Genome Diagnostics can be found at <https://www.radboudumc.nl/en/afdelinggen/genetica/about-us/genomediagnosics>

Service	Turnaround time	Required material
Exome sequencing diagnostics (WES)	Exome gene panel analysis: 2-3 months	Preferably: - 2 x 3-6ml EDTA blood - 20 ml amniotic fluid (only Rapid) or >30 ml (Rapid + QF-PCR growing cells)
	Exome gene panel analysis followed by exome wide analysis (in one report): 2-3 months	If there is no blood and/or amniotic fluid available: - Desired amount DNA: 5ug - Absolute minimum: 3ug (minimum concentration is 25 ng/ul, volume 30 ul). Prenatal material minimum is 450 ng (30 ul of 15 ng/ul)
	Rapid Trio/de novo analysis of proband and both parents: <15 business days	
Interpretation of exome data	2 months	n/a
Array diagnostics (genome wide)	5 weeks	2 x 3-6ml EDTA blood
Multiple gene diagnostics (Gene panels)	3-8 weeks*	2 x 3-6ml EDTA blood
Single gene diagnostics	4-8 weeks*	2 x 3-6ml EDTA blood
Mutation diagnostics (carrier testing)	4 weeks	Gene/array diagnostics: 2 x 3-6ml EDTA blood Chromosome diagnostics (karyotyping): 2 x 5ml Heparin blood in Natrium- or Lithium-Heparin tubes (neonates 1-2ml)
Pharmacogenetics	1-8 weeks*	2 x 3-6ml EDTA blood
Chromosome diagnostics	2-5 weeks	Karyotyping: 2 x 5ml Heparin blood in Natrium- or Lithium-Heparin tubes (neonates 1-2ml) QF-PCR: neonates: 1-2ml EDTA blood
FISH	2-5 weeks	2 x 5ml Heparin blood in natrium- of lithium-heparin tubes
Urgent request	Please, contact: gen@radboudumc.nl or Tel: +310243613799, TAT depends on technique	Please, see material under requested service
mtDNA	4-12 weeks	Cooled (none frozen) urine, we prefer 50-100ml

* In our application system you will find the exact turnaround times for each individual test